Saskatchewan
Research Council

Incidence Of 1;29 Robertsonian Translocation In Canadian Beef Cattle

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SRC Technical Report No. 231

Publication S-803-11-B-91

January, 1991

91-07198/

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INTRODUCTION

Chromosomes contain the genetic material of living organisms. The normal chromosome complement of cattle, *Bos taurus*, is 60: 58 (29 pairs) acrocentric autosomes, and 2 (XX = female, XY= male) sex chromosomes (Fig. 1). The sex chromosomes can readily be identified by their metacentric centromeres; the X is a large and the Y is a very small submetacentric chromosome (Gustavsson, 1969).

Occasionally, chromosomal rearrangements occur in individual animals. Centric fusion or Robertsonian translocation, named after W. R. B. Robertson, results from the joining of 2 acrocentric chromosomes at or around the centromere, forming a large metacentric or submetacentric chromosome (Robertson, 1916).

The 1;29 Robertsonian translocation in cattle was first discovered by Gustavsson and Rockborn (1964). In this particular translocation, chromosome no. 1 is fused to chromosome no. 29, and a reduction in the total chromosome number always occurs. Individuals heterozygous or homozygous for the 1;29 Robertsonian translocation have 59 and 58 chromosomes, respectively, instead of a diploid number of 60 (Gustavsson, 1979) (Fig. 2).

Table 1.
Various cattle breeds carrying the 1;29 Robertsonian translocation.

Baoulé	Limousin
Black Pied	Marchigiana
Blonde d'Aquitaine	Montybéliard
British White	Ottonese
Brown Atlas	Pisana
Brown Mountain	Podolian
Charolais	Polish Red
Chianina	Red Pied
Fighting Bull (De Lidia)	Red Poll
Gascony	Romagnola
German Red Pied	Santa Gertrudis
Grauvieh	Thai (Siamese)
Guernsey	Simmental
Holstein Friesian	Swedish Red and White
Japanese Black	Vosges

Data from Gustavsson (1979) and Long (1985)

The 1;29 translocation has been reported in numerous breeds of cattle worldwide (Gustavsson, 1979; Long, 1985) (Table 1). This translocation is primarily found among European continental beef breeds and Scandinavian dairy breeds, but occurs very rarely in British beef breeds and the Holstein Friesian cattle (McWhir and Church, 1987). Whether the Robertsonian translocation in different geographical areas and breeds is due to recurrent mutation or distribution of an ancient mutation is not known (Gustavsson, 1979).

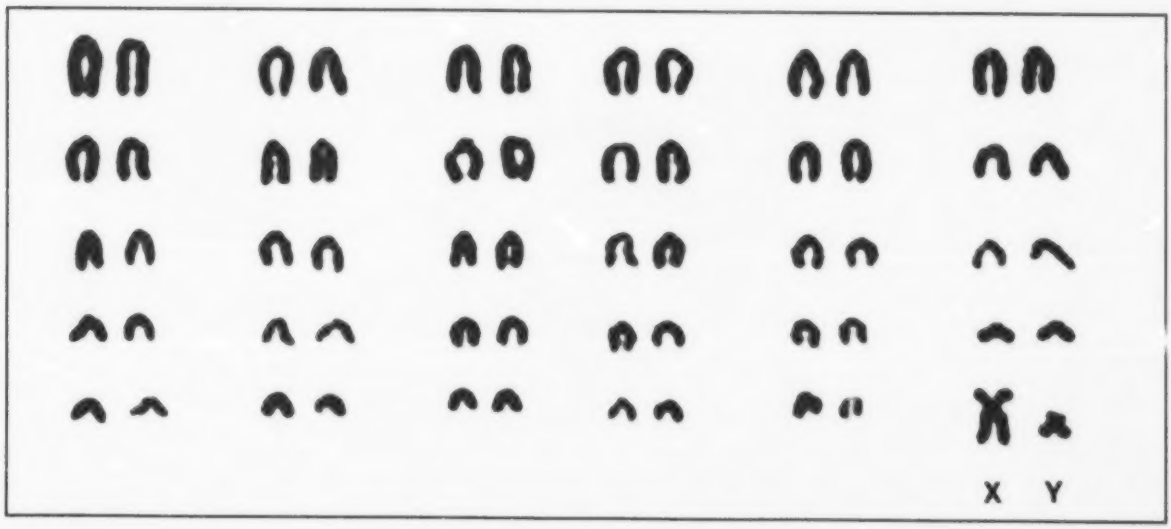

Fig. 1. Normal cattle (*Bos taurus*) karyotype consisting of 58 acrocentric autosomes and 2 submetacentric sex chromosomes; XY=male.

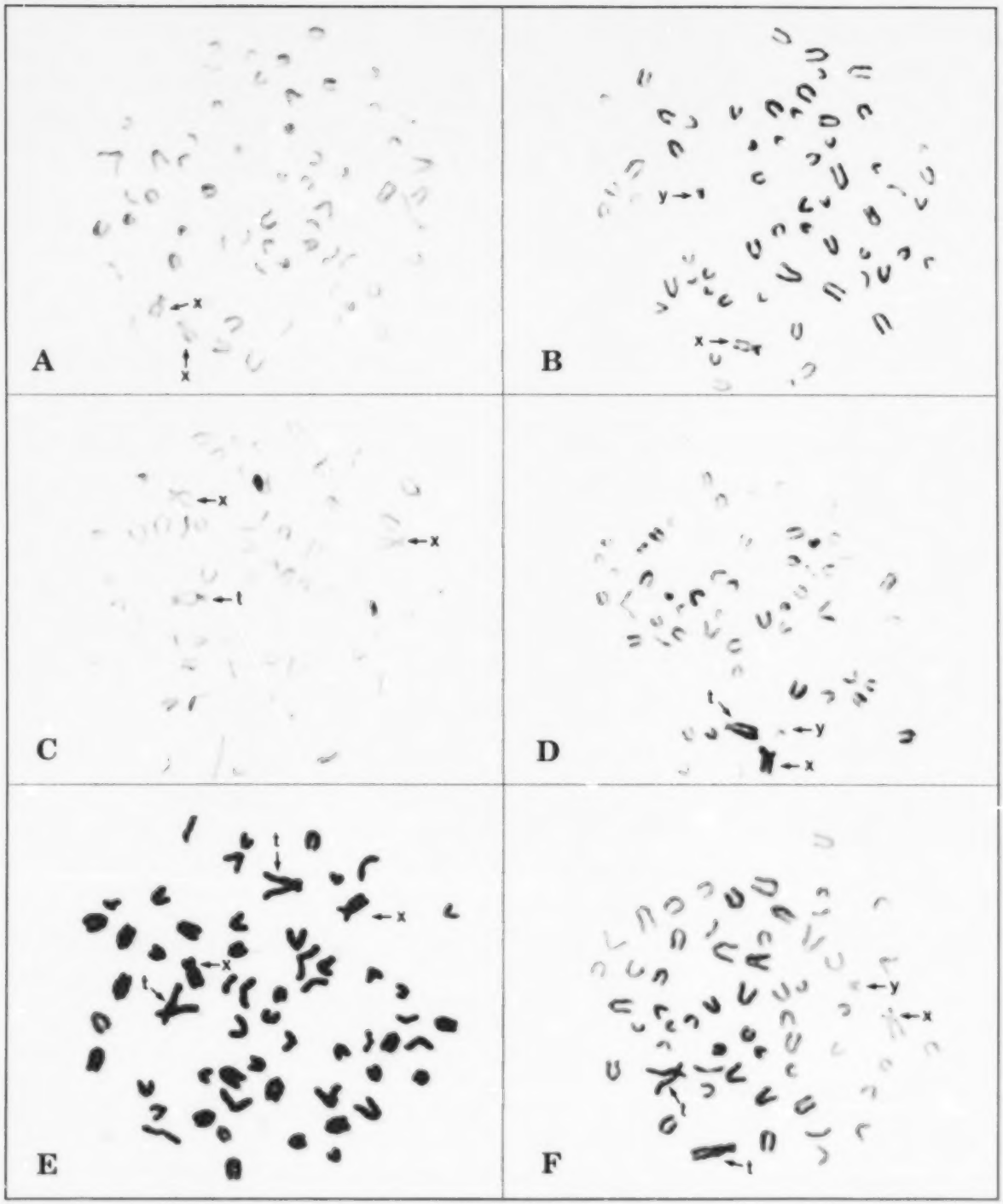

Fig. 2. Cultured leucocyte mitotic chromosomes from: A: normal female, 60,XX; B: normal male, 60,XY; C: female heterozygous for the 1;29 translocation, 59,XX,t; D: male heterozygous for the 1;29 translocation, 59,XY,t; E: female homozygous for the 1;29 translocation, 58,XX,tt; F: male homozygous for the 1;29 translocation, 58,XY,tt. Sex chromosomes are designated by letters X and Y, and translocations are indicated by the letter t.

LITERATURE REVIEW

Chromosomal aberrations are apparently responsible for a high percentage of embryonic deaths in mammals (Fechheimer, 1971; Schmutz et al., 1990a). The effects of genetic abnormalities largely depend on the amount of genetic material added or lost. In some abnormalities, like the 1;29 Robertsonian translocation, chromosomes are merely altered in morphology without a significant loss of genetic material, thus the genetic makeup remains balanced. Balanced rearrangements cause no apparent phenotypic abnormality, and therefore bovine carriers of the 1;29 Robertsonian translocation appear to be phenotypically normal (McWhir and Church, 1987). Despite their normal morphological features, bulls heterozygous for the 1;29 Robertsonian translocation exhibit reduced fertility by approximately 5 - 10% (Dryendahl and Gustavsson, 1979; Gustavsson, 1979; Logue and Harvey, 1978; Long, 1985; Weber et al., 1987). In natural service, heterozygous carrier bulls sired fewer calves than karyotypically normal bulls (McWhir and Church, 1987). These animals, however, show normal mating behavior, libido, semen quality, spermatogenesis, and body conformation (Gustavsson, 1969; Logue and Harvey, 1978).

Females heterozygous for the 1;29 Robertsonian translocation also exhibit lower fertility when compared to control animals (Gustavsson, 1969, 1971b; Maurer and Vogt, 1988; Refsdal, 1976; Schmutz et al., 1990b). Gustavsson (1971a) and Pinheiro et al. (1987) found approximately 31 and 25%, respectively, of repeat breeder heifers carried the 1;29 translocation. Studies by King and Linares (1983) and Swartz and Vogt (1983) indicate approximately 13 - 18% of chromosomal abnormalities are found among subfertile heifers. When compared to noncarrier females, 1;29 Robertsonian translocation carriers showed significantly lower calving efficiency (Maurer and Vogt, 1988). In one

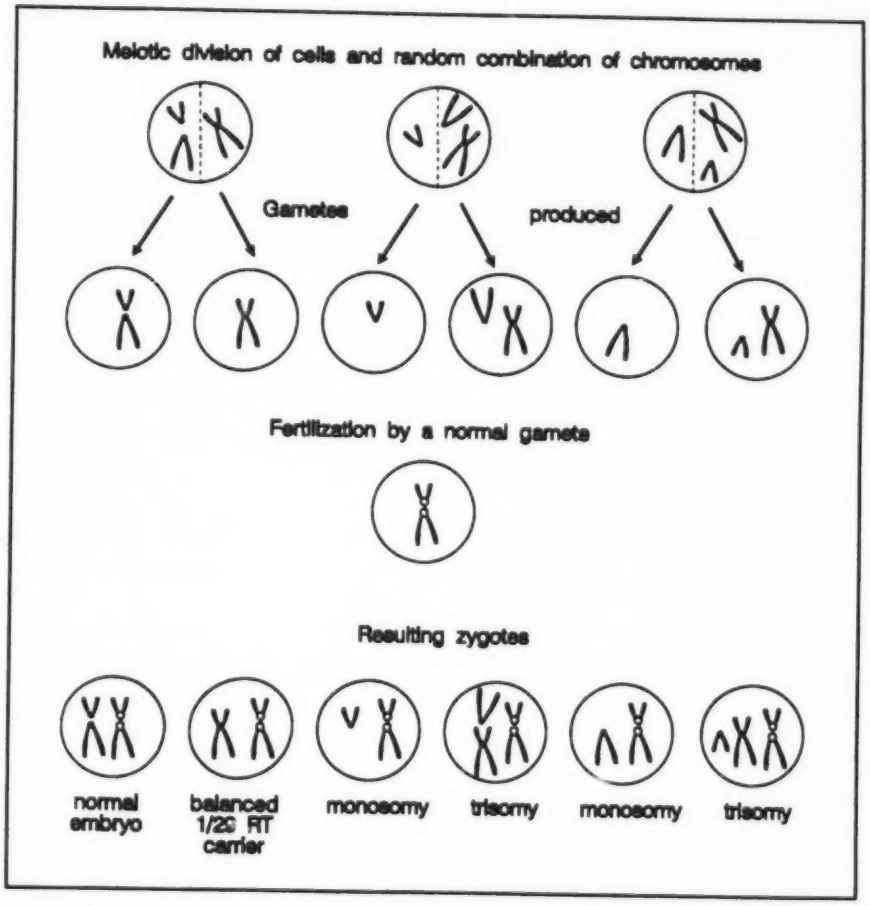

Fig. 3. Diagram depicting balanced segregation and non-disjunction at meiosis: resulting zygotes or embryos may be normal, balanced 1;29 Robertsonian translocation carriers, monosomic and trisomic.

Charolais herd, 2 out of 3 1;29 Robertsonian translocation carrier cows aborted calves, and 2 out of 3 delivered stillborn calves (Schmutz and Moker, 1989).

In structural chromosomal abnormalities, like the 1;29 Robertsonian translocation, fertility is impaired since meiotic segregation leads to the production of unbalanced gametes (Cribiu et al., 1989; Long, 1985). Cattle heterozygous for the 1;29 translocation have an uneven number of chromosomes, 59 instead of 60. At meiosis, 2 acrocentric homologues and the fused 1;29 chromosome form a trivalent. If balanced segregation occurs, then fertilization by normal gametes produces normal zygotes and balanced translocation carrier zygotes. On the other hand, in nondisjunction, fertilization by normal gametes produces zygotes which may be missing 1 chromosome (monosomy), or contain an extra chromosome (trisomy) (Fig. 3).

Popescue (1980) and King et al. (1980) studied embryos fertilized by semen from bulls heterozygous for the 1;29 Robertsonian translocation. Popescue (1980) noted that 2/52 embryos with unbalanced karyotypes appeared to be monosomic for chromosome no. 1. Out of 18 embryos analyzed by King et al. (1980) 9 were balanced for the translocation, while 2 embryos appeared to be trisomic for chromosome no. 1. Both monosomy and trisomy are usually lethal, resulting in early embryonic death (Coates et al., 1988; Gustavsson, 1969; Long, 1985; Refsdal, 1976; Schmutz et al., 1990b). Nondisjunction of 2.8% was found in normal animals compared to 6.4% in heterozygous 1;29 Robertsonian translocation carrier animals (Logue and Harvey, 1978).

A major decrease in fertility among 1;29 translocation carrier cows appears to be largely due to formation of unbalanced embryos. Carrier females show somewhat greater reproductive impairment than carrier bulls: 30% of embryos revealed an unbalanced karyotype when the cow was the 1;29 translocation carrier compared to 20% when the bull carried the translocation (Schmutz et al., 1990b). In addition, superovulation experiments indicate that 1;29 Robertsonian translocation carrier cows produce on average fewer embryos than chromosomally normal cows (Schmutz et al., 1990b). Failure to conceive and early embryonic deaths reduce pregnancy rates, increase the age at conception, and eventually lead to carriers being culled as open females or repeat breeders (Maurer and Vogt, 1988). There are no differences, however, in body conformation, meat, milk, and butterfat production between carrier and noncarrier cows (Gustavsson, 1969, 1979).

The 1;29 Robertsonian translocation appears to have a worldwide distribution (Gustavsson, 1979). The seriousness of this translocation is that it is quite common in cattle, is associated with reduced fertility, and is passed on from one generation to the next (Swartz and Vogt, 1983). A heterozygous 1;29 translocation carrier mated to a normal noncarrier will produce offspring of which 50% will be carriers and 50% noncarriers (McWhir and Church, 1987; Schmutz and Moker, 1989; Weber et al., 1989). When a cow and bull, both carriers of the 1;29 translocation are mated, normal as well as heterozygous and homozygous translocation carrier calves will be born. It is predicted, however, that the number of aborted embryos (monosomic and trisomic) would substantially increase (Schmutz et al., 1990c). When animals homozygous for the 1;29 Robertsonian translocation are bred, theoretically, all offspring will carry the translocation but unbalanced gametes should not be produced (Schmutz et al., 1990a).

Frequent use of heterozygous or homozygous 1;29 Robertsonian translocation carrier bulls through natural service will rapidly disperse the translocation throughout a herd. Increased use of artificial insemination and embryo transfer will also quicken the spread of the translocation, which may further result in serious consequences from undetected chromosomal abnormalities (Buoen et al., 1988; Cribiu et al., 1989; Schmutz and Moker, 1989; Swartz and Vogt, 1983; Weber et al., 1989).

MATERIALS AND METHODS

Seventy-five purebred cattle from Canadian beef producers were karyotyped upon request at the Bovine Blood Typing Laboratory of the Saskatchewan Research Council. Blood samples were collected in heparinized, sterile, 10 ml vacutainers. Cell cultures were set up as soon as possible after collection using 10 ml of Ham's F-10 medium, containing 16% fetal calf serum, fungizone, and gentamycin, 20 drops of whole blood, and 0.2 ml of phytohemaglutinin. Cultures were incubated for 2 or 3 days at 37° -38°C. Fifty minutes before harvesting, 0.1 ml of colcemid was added to arrest cells at metaphase. The leucocytes were then suspended in 0.075 M KCl hypotonic solution for 20 min. and fixed in freshly prepared Carnoy's solution (3:1 methanol:glacial acetic acid) overnight. The following day cells were washed 2 to 3 times in fresh Carnoy's solution, spread on glass slides, and stained with Giemsa stain. Microscopic chromosomal analysis was performed on a minimum of 15 cells from each individual to rule out mosaicism at the 20% level with 95% confidence (Hock, 1977).

Table 2.
Karyotypes of male and female cattle examined.

Description	Karyotype	No.
Normal male	60,XY	52
Normal female	60,XX	8
Male heterozygous for 1;29 Robertsonian translocation	59,XY,t	11
Female heterozygous for 1;29 Robertsonian translocation	59,XX,t	1
Male homozygous for 1;29 Robertsonian translocation	58,XY,tt	2
Female homozygous for 1;29 Robertsonian translocation	58,XX,tt	0
Chimerism	60,XY/60,XX	1

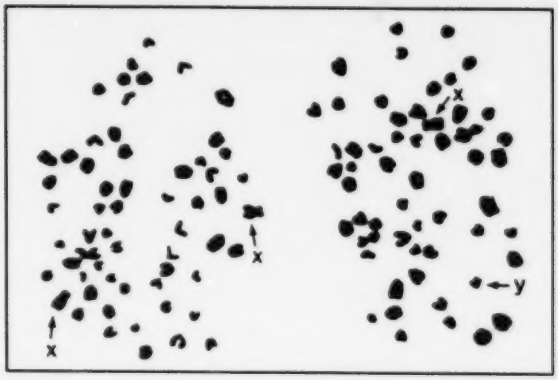

Fig. 4. Mitotic chromosome spreads showing leucocyte chimerism.

RESULTS AND DISCUSSION

Blood samples from 66 males and 9 females from Blonde d'Aquitaine, Charolais, Hereford, Limousin, and Simmental breeds were karyotyped. Fifty-two males and 8 females had a normal karyotype, 11 males and 1 female were heterozygous, and 2 males were homozygous for the 1;29 Robertsonian translocation (Table 2). In addition the karyotype of 1 bull showed the presence of both XX and XY pex chrom, known as leucocyte chimerism (Fig. 4). Twenty percent of the cattle tested had abnormal karyotypes, and 19% were carriers of the 1;29 translocation.

The 1;29 Robertsonian translocation is definitely present in Canadian beef cattle, but the percentage of cattle carrying this translocation varies among different breeds as well as among herds. It should be noted, however, that the cattle examined in this study were not a random sample, and therefore the high percentage of abnormal karyotypes is not an accurate estimate of the spread of the 1;29 Robertsonian translocation in Canadian cattle. In studies by McWhir et al. (1987) and Schmutz et al. (1990a), 3.5% (9/253) and 5% (7/133) of Canadian beef cattle, respectively, exhibited the 1;29 centric fusion.

In our study, 16% of the cattle tested had

one copy (2n =59) of the translocation. Research shows that despite their physically normal appearance different breeds of cattle heterozygous for the 1;29 translocation exhibit some decrease in fertility (Gustavsson, 1979; Maurer and Vogt, 1988; McWhir and Church, 1987; Schmutz and Moker, 1989). Carrier bulls may sire fewer calves, while carrier females exhibit prolonged calving intervals, repeat breeding, reduced calving efficiency, calving difficulty, abortion and stillbirth, all resulting in lower fertility and reproductive wastage (Coates et al., 1988; Dryendahl and Gustavsson, 1979; Gustavsson, 1971; Maurer and Echternkamp, 1985; Maurer and Vogt, 1988; Refsdal, 1976; Schmutz et al., 1990a, 1990b).

Males and females homozygous for the 1;29 Robertsonian translocation have also been diagnosed. Bulls with a chromosomal complement of 2n=58 have been used in AI centers, have sired calves (Gustavsson, 1969), and are capable of producing viable sperm (Schmutz and Moker, 1989). In our study, 2/75 animals (3%) were bulls with 2 copies of the 1;29 translocation, but we have no information regarding their fertility or lack thereof. Females homozygous for the 1;29 translocation are also capable of producing offspring (Schmutz and Moker, 1989). Our small sample size did not include any females with 2 copies of the 1;29 centric fusion. The limited data available indicates that cattle of both sexes with 2 copies of the 1;29 Robertsonian translocation are not sterile, but the degree of fertility or subfertility is not yet known (Schmutz et al., 1990a).

Karyotyping positively identifies chromosomal abnormalities. Unfortunately, no specific physical trait or marker has been found to distinguish 1;29 Robertsonian translocation carriers from chromosomally normal animals. Small scrotal size does not necessarily denote the 1;29 centric fusion, nor is there any association between the presence or absence of horns and an abnormal karyotype (Schmutz, 1989; Schmutz et al., 1990a). Selective breeding can quickly eliminate genetic abnormalities associated with morphological defects, but translocations which do not exhibit visible phenotypic changes are not as easy to eliminate by selective breeding (Cribiu et al., 1989).

SUMMARY

Our results show that the 1;29 Robertsonian translocation is present in Canadian purebred beef cattle. Unfortunately, there is no definite legislation in North America regarding translocations (Schmutz and Moker, 1989). Elsewhere, much international concern exists over the effects of chromosomal abnormalities. Good reproductive performance is of considerable importance in the livestock industry. Keeping cattle under cytogenetic control is economically advantageous since high fertility results in numerous offspring which translates into bigger profits in both meat and milk production. Cytogenetic investigations are performed routinely in France, Great Britain, Hungary, West Germany, and Norway (Gustavsson, 1979). Many countries now demand that imported semen and breeding animals be free of chromosomal abnormalities: Australia and Great Britain require cytogenetic testing of all imported cattle (Buoen et al., 1988). In Sweden, the Swedish Red and White breed was subjected to an eradication program where all bulls carrying the 1;29 Robertsonian translocation were eliminated from AI. The result of the eradication was an increase in fertility for the whole Swedish Red and White cattle population (Gustavsson, 1979).

Canadian breed associations should become more aware of the presence and problems associated with chromosomal abnormalities in their specific breeds. New policies may be needed to decrease or even eliminate Robertsonian translocations in AI bulls and affected herds. Without cytogenetic testing, undetected chromosomal abnormalities may cause a serious reduction in potential productivity, and consequently result in economic losses to beef producers.

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